

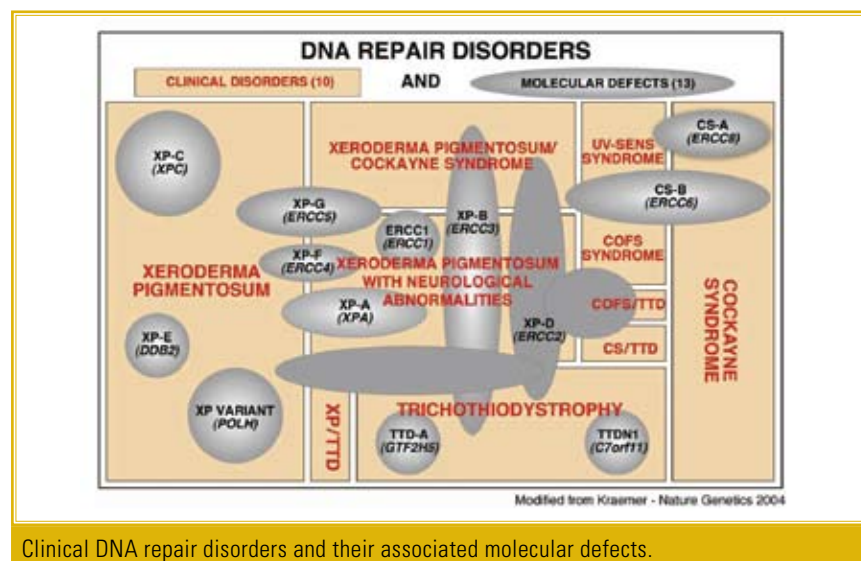
# A Look at Rare Diseases, from Molecules to Patients

*Workshop on Xeroderma pigmentosum, Cockayne syndrome, and trichothiodystrophy examines diseases of DNA repair, cancer, and premature aging.*

Most people do not welcome the signs of aging, but for patients with Xeroderma pigmentosum (XP) and related diseases, the issue is not one of social status but of survival. XP causes patients to develop life-threatening skin cancers and a prematurely aged appearance of sun-exposed skin due to the inability to repair cell damage by ultraviolet (UV) radiation. There are other related disorders such as Cockayne syndrome (CS) and trichothiodystrophy (TTD) that also produce premature aging and are caused by inherited mutations that alter multifunctional protein complexes, which play essential roles in DNA repair and RNA transcription.

These rare diseases have a combined worldwide incidence estimated at less than one per 250,000 births. Yet, there are enough dedicated researchers and clinicians interested in XP, CS, and TTD, and enough cooperative patients and family members, that remarkable progress has been made recently in understanding the molecular basis of these complex disorders. This progress was evident at a workshop to address these rare diseases called Xeroderma Pigmentosum and Other Diseases of Human Premature Aging and DNA Repair: Molecules to Patients held in Chantilly, VA from September 21-24, 2010.

One hundred researchers, clinicians, affected patients, and



Clinical DNA repair disorders and their associated molecular defects.

representatives of patient support groups gathered at the workshop—co-organized by Kenneth Kraemer, M.D., Senior Investigator in CCR's Dermatology Branch; Vilhelm Bohr, M.D., Ph.D., Chief of the National Institute of Aging's Laboratory of Molecular Gerontology; and Laura Niedernhofer, M.D., Ph.D., Associate Professor at the University of Pittsburgh—to share, consider, and discuss the latest developments in understanding XP and other human diseases characterized by cancer, premature aging, and defects in DNA repair.

The third in a series, this workshop emphasized discussion, interaction, and open exchange of information and ideas among bench scientists, clinicians, patients, and patient advocates in order to establish new collaborations. The workshop sessions covered a variety of topics on XP, CS, and TTD including natural history and clinical features of disease, clinical and laboratory diagnosis, therapeutic approaches, molecular analysis of accelerated aging, neurodegeneration in Huntington's disease, as well as DNA repair and genome instability.

The meeting revealed areas in which great progress has been

made, areas ripe for future study, and bottlenecks that are inhibiting progress in either basic understanding of the diseases or their clinical management. Several panel discussions and poster sessions provided the unique opportunity for extensive conversation and interaction between clinicians, researchers, patients, and family support group members in both formal and informal settings.

"The presence and participation of patients, their advocates, and family support groups was an important and enriching feature of this and the preceding two workshops in this series, in 2004 and 2006," noted Dr. Kraemer. "It is important to emphasize that studying relatively rare diseases such as XP, CS, and TTD may lead to insights that are relevant for more common diseases such as cancer and neurodegeneration. For example, XP patients have a 10,000-fold risk of developing new skin cancers and melanomas." The efforts of all the participants at this workshop may contribute to a greater understanding of rare diseases, as well as a better insight into the risk factors for common diseases in the general population.

(Image: K. Kraemer, CCR)